

ABSTRACT OF THE DISCLOSURE

A method for production of rhPBGD in high scale and use of rhPBGD in a method for treatment or prophylaxis of disease caused by deficiency, in a subject, of an enzyme belonging to the heme biosynthetic pathway. The method comprising administering, to the subject, an effective amount of one or more catalyst which is said enzyme or an enzymatically equivalent part or analogue thereof preferable in combination with a gene therapy of a mutation related to the catalyst. The disease is selected from the group consisting of acute intermittent porphyria (AIP, ALA deficiency porphyria (ADP), Porphyria cutanea tarda (PCT), Hereditary coproporphyria (HCP), Hardeorporphyria (HDP), Variegata porphyria (VP), Congenital erythropoietic porphyria (CEP), Erythropoietic protoporphyria (EPP), and Hepatoerythropoietic porphyria (HEP). The catalyst is one or more enzymes selected from the group consisting of delta-aminolevulinic acid synthetase, delta-aminovulnic acid dehydratase (ALAD), porphobilinogen deaminase (PBGD), uroporphyrinogen III cosynthetase, uroporphyrinogen decarboxylase, coproporphyrinogen oxidase, protoporphyrinogen oxidase, and ferrochelatase, or an enzymatically equivalent part or analogue thereof. In addition the invention relates to the use of rhPBGD. The invention also relates to an expression plasmid pExpl-M2-BB (seq. ID No.1) and to use of a DNA fragment, the EcoR 1-Hind III linear fragment (seq. ID No. 2), used for transformation in the hemC disruption strategy for production of rhPBGD expressed in *E.coli*.